



NOTES

T-CELL DEFICIENCIES

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Impaired function of T lymphocytes due to thymic hypoplasia → deficiency of cell-mediated immunity → high risk of infections
- Two main causes of thymic hypoplasia
 - DiGeorge syndrome (DGS)/22q11.2 deletion syndrome
 - Ataxia-telangiectasia (AT) syndrome (see Chapter 84 in Volume 1)
- Inherited, due to genetic defects
- Associated with other congenital abnormalities

SIGNS & SYMPTOMS

- Recurrent infections; usually viral
- Depend on cause of thymic hypoplasia

DIAGNOSIS

LAB RESULTS

Blood tests

- T cell numbers, function
- Calcium levels
 - Hypocalcemia → DGS, eucalcemia → AT

OTHER DIAGNOSTICS

- Genetic testing
 - Identify genetic defect

TREATMENT

MEDICATIONS

- Management of symptoms
 - Antibiotic therapy/prophylaxis for infections

SURGERY

- Thymus transplantation

VISIT...

LANZAROTE
Caliente.COM

DIGEORGE SYNDROME (DGS)

osms.it/digeorge-syndrome

PATHOLOGY & CAUSES

- AKA 22q11.2 deletion syndrome
- Genetic condition; q11.2 portion of DNA on chromosome 22 deleted → deletion of TBX gene on chromosome 22 → **impaired development of pharyngeal pouches 3, 4** → hypoplasia of thymus, inferior parathyroid gland
 - **Deficiency** in mature **T cells**, adaptive immune response: complete thymic aplasia can be fatal in first year of life
 - Parathyroid hypoplasia → low levels of parathyroid hormone → **hypocalcemia**
- 22q11.2 region encodes genes that affect other organs/tissues → congenital **cardiac defects**; facial, developmental abnormalities; behavioral, mental health conditions

SIGNS & SYMPTOMS

- **Characteristic facial appearance**: long face, small teeth, broad nose
- Developmental abnormalities (e.g. cleft palate)
- Cardiac defects (e.g. **truncus arteriosus**, **tetralogy of Fallot**)
- Thymus gland abnormalities → underdevelopment of cellular immune system → susceptibility to **recurrent** sinopulmonary **infections**/severe combined immunodeficiency (SCID)
- Learning difficulties/mental health conditions (e.g. schizophrenia)
- Hypoparathyroidism → hypocalcemia → seizures, tetany, osteoporosis

DIAGNOSIS

LAB RESULTS

Blood tests

- Reduced T cell number, function
- Hypocalcemia
- Low levels of **parathyroid hormone**

OTHER DIAGNOSTICS

- Genetic testing

TREATMENT

MEDICATIONS

- Management of symptoms
 - Antibiotics for infections

SURGERY

- Thymus transplantation

OTHER INTERVENTIONS

- Vitamin D, calcium supplements for hypocalcemia



Figure 35.1 A child with DiGeorge syndrome.